

ISCN

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EXHIBIT

E

46,X,r(X).ish r(X)(p22.3q21)(KAL+,DXZ1+,XIST+,DXZ4-)

A ring X was further defined by ish as containing the short arm marker KAL, the X alpha-satellite DXZ1 and the XIST gene on the long arm. It does not include DXZ4 at Xq24.

46,X,+r.ish r(X)(wcpX+,DXZ1+)

A ring chromosome was identified by ish as a derivative X chromosome using whole chromosome paint X and X alpha-satellite probe DXZ1.

46,X,?i(Y)(p10).ish idic(Y)(q11)(DYZ3++,DYZ1-)

A presumed isochromosome for the short arm of Y was shown by ish to have two centromeres and no heterochromatin.

45,XY,der(14;21)(q10;q10).ish dic(14;21)(p11.2;p11.2)(D14Z1/D22Z1+,D13Z1/D21Z1+)

A Robertsonian translocation, rob(14;21) or der(14;21), shown to be dicentric using ish.

46,XX.ish inv(16)(p13q22)(pcp16q sp)

A normal female chromosome complement on routine cytogenetic analysis was found to have a pericentric inversion of chromosome 16 by ish utilizing a partial chromosome paint for the long arm of chromosome 16 that was split by the inversion.

46,XY,inv(17)(p13q21).ish inv(17)(p13.1q21.3)(D17S379 st,RARA inv)

An inverted chromosome 17 was further defined by ish using probes for D17S379 (which stayed stationary) and RARA (which moved relative to D17S379).

46,XX.ish t(4;11)(p16.3;p15)(wcp11+,D4F26-,D4S96+,D4Z1+;D4F26+,wcp11+)

A cryptic reciprocal translocation between chromosomes 4 and 11 was identified by ish. The der(4) was positive with whole chromosome paint 11, a probe for D4S96 (Wolf-Hirschhorn region) and 4 alpha-satellite but negative for D4F26 (4p telomere region). The der(11) was positive for D4F26 as well as whole chromosome paint 11.

46,XX.ish der(4)t(4;11)(p16.3;p15)mat(wcp11+,D4F26-,D4S96+,D4Z1+)

This child is an unbalanced offspring from the segregation of the cryptic translocation above. She has one normal chromosome 4 and two normal chromosomes 11. The ish results of the der(4) are the same as der(4) of the mother.

46,XY.ish 4(D4F26x2,D4S96x2)

A normal male (father of the child in the previous example) was tested by ish using probes for loci D4F26 and D4S96. There were two copies of both.

46,XX,ins(2)(p13q21q31).ish 2(wcp2+)

A direct insertion of the long arm segment 2q21-q31 into the short arm at band 2p13 was confirmed as derived from chromosome 2 by ish using whole chromosome paint 2.

46,XY,ins(5;2)(p14;q32q22).ish ins(5;2)(wcp2+;wcp2+)

An inverted insertion of a chromosome 2 segment into the short arm of chromosome 5 was confirmed as derived from chromosome 2 using whole chromosome paint 2.